**SOUTHERN RIVER COLLEGE**

**Human Biology**

**Unit 4**

**TASK 7**

**Mutations & Gene Pools Validation (5%)**

**TYPE:** Extended Response

**CONTENT:** Mutations & Gene Pools

You are to complete the following questions, using your knowledge gained from the take home booklet. You are allowed Part A ONLY as notes during this Validation.

Take home booklet / 3 marks

References / 2 marks

Validation / 25 marks

**TOTAL / 30 marks**

**\_\_\_\_\_\_%**

*Use the information below to help you answer question 1, 2 and 3.*

It is thought that man evolved in Africa and then migrated from there to populate the world. The table below shows blood group percentages in various ethnic groups. The geographical locations of these populations can be found on the following page, using the map references.

**TABLE 1:**

|  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- |
|  | **Map reference** | **O** | **A** | **B** | **AB** |
| **Australian Aboriginal** | A | 61 | 39 | 0 | 0 |
| **English** | B | 47 | 42 | 8 | 3 |
| **Eskimos**  **(Alaska)** | C | 38 | 44 | 13 | 5 |
| **Eskimos (Greenland)** | D | 54 | 36 | 23 | 8 |
| **USA (black)** | E | 49 | 27 | 20 | 4 |
| **USA (white)** | F | 45 | 40 | 11 | 4 |
| **Peru (Indians)** | G | 100 | 0 | 0 | 0 |
| **Navajo (Native American Indian)** | H | 73 | 27 | 0 | 0 |



E & F

H

G

D

C

B

A

1. Suggest why Peruvian Indians and Aborigines have evolved with no incidence of B or AB blood groups in their populations.

(2 marks)

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1. If the B or AB blood group were to appear in the Aboriginal population, how could this be explained?

(2 marks)

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1. Identify and highlight the location of the Eskimo (Alaskan population), Navajo and Peruvian Indians on the map provided.
2. What can the blood group data from Table 1, for the Eskimo (Alaskan population), Navajo and Peruvian Indians tell us about the spread of people through the Americas’ populations? (3 marks)

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*Use the information below to answer question 4.*

The Andaman and Nicobar islands are located southeast of the **Indian subcontinent**, separated by the **Bay of Bengal** by about 1,300 km. These islands are outlined by the black box.

Table 2 shows the blood group percentages for the populations on these islands (Grand Andamanese and Nicobarese) and those people on the Indian mainland.



|  |  |  |  |  |
| --- | --- | --- | --- | --- |
| **Blood Group** | **O** | **A** | **B** | **AB** |
| **Grand Andamanese (Andaman island)** | 9 | 60 | 23 | 9 |
| **Nicobarese (Nicobar island))** | 74 | 9 | 15 | 1 |
| **Indians (India – General population)** | 37 | 22 | 33 | 7 |

**TABLE 2:**

1. In December 2004 a massive earthquake in the Indian Ocean resulted in a tsunami that swept across these islands and coastal India. This may have led to a ‘bottleneck effect” where only a few survive in certain populations.
   1. Explain what effect the bottleneck may have on blood groups percentages present in the Grand Andamanese and Nicobarese populations.   
        
      **Use the data in the table to support your answer**. (6 marks)

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* 1. From the information given, name an evolutionary process that doesn’t appear to be influencing the allele frequencies of the populations.

(1 mark)

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* 1. Describe how the process you mentioned in Question 4b) affects the genotype frequencies of populations in general.

(2 marks)

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1. For the diseases listed below, describe the effect they have on the population, the processes that maintain them in the population AND why it may be of benefit:
   1. Sickle cell anaemia (3 marks)

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* 1. Thalassemia (3 marks)

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* 1. Tay Sach’s Disease (chronic form) (3 marks)

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**SOUTHERN RIVER COLLEGE**

**Human Biology**

**Unit 4**

**TASK 7**

**Mutations & Gene Pools MARKING KEY (5%)**

**TYPE:** Extended Response

**CONTENT:** Mutations & Gene Pools

You are to complete the following questions, using your knowledge gained from the take home booklet.

Take home booklet / 3 marks

References / 2 marks

Validation / 25 marks

**TOTAL / 30 marks**

**\_\_\_\_\_\_%**

*Use the information below to help you answer question 1, 2 and 3.*

It is thought that man evolved in Africa and then migrated from there to populate the world. The table below shows blood group percentages in various ethnic groups.

**TABLE 1:**

|  |  |  |  |  |
| --- | --- | --- | --- | --- |
|  | **O** | **A** | **B** | **AB** |
| **Australian Aboriginal** | 61 | 39 | 0 | 0 |
| **English** | 47 | 42 | 9 | 3 |
| **Eskimos**  **(Alaska)** | 38 | 44 | 13 | 5 |
| **Eskimos (Greenland)** | 54 | 36 | 23 | 8 |
| **USA (black)** | 49 | 27 | 20 | 4 |
| **USA (white)** | 45 | 40 | 11 | 4 |
| **Peru (Indians)** | 100 | 0 | 0 | 0 |
| **Navajo (Native American Indian)** | 73 | 27 | 0 | 0 |

1. Suggest why Peruvian Indians and Aborigines have evolved with no incidence of B or AB blood groups in their populations.
2. marks)

*1 Founder effect / genetic drift*

*1 No ‘B’ alleles present in original migrants*

1. If the B or AB blood group were to appear in the Aboriginal population, how could this be explained?

(2 marks)

*1 Migration*

*1 someone with ‘B’ allele has arrived in the population and has passed on through mating*

*OR*

*1 Random mutation*

*1 Either ‘O’ or ‘A’ allele has had a mutation that mimics ‘B’ antigen*

2. What can the data for the Eskimo (Alaskan population), Navajo and Peruvian Indians tell us about the spread of people through the Americas’ populations?

(3 marks)

*Any 3*

*1 Not a consistent migration*

*1 probably started in Alaska (all alleles present)*

*1 moved south in stages*

*1 Navajo is founder as loss of alleles is gradual*

*1 Peru has clear founder effect as 2 alleles lost*

The Andamen and Nicobar islands are located southeast of the [Indian subcontinent](http://en.wikipedia.org/wiki/Indian_subcontinent), separated by the [Bay of Bengal](http://en.wikipedia.org/wiki/Bay_of_Bengal) by about 1,300 km.



|  |  |  |  |  |
| --- | --- | --- | --- | --- |
| **Blood Group** | **O** | **A** | **B** | **AB** |
| **Grand Andamanese** | 9 | 60 | 23 | 9 |
| **Nicobarese (Nicobars)** | 74 | 9 | 15 | 1 |
| **Indians (India - General)** | 37 | 22 | 33 | 7 |

**TABLE 2:**

1. In December 2004 a massive earthquake in the Indian Ocean resulted in a tsunami that swept across these islands and coastal India. This may have led to a ‘bottleneck effect” where only a few survive in certain populations.
   1. Explain what effect the bottleneck may have on blood groups percentages present in the Grand Andamanese and Nicobarese populations.   
        
      **Use the data in the table to support your answer**. (6 marks)

*ANY 3 of the following: at least 1 mark must include data.*

*Andamananese*

*1 most likely loss of ‘O’ allele*

*1 as only 9% of population carry it*

*1 ‘A’ allele will increase further*

*1 as 60% carry it*

*1 AB blood group could increase*

*1 A and B individuals producing offspring of AB blood group*

*ANY 3 of the following: at least 1 mark must include data.*

*Nicobarese*

*1 loss of ‘A’ alleles, possibly ‘B’ too*

*1 as only 9% and 15% respectively remain*

*1 increase in ‘O’ allele*

1. *as already 74%*
   1. From the information given, name an evolutionary process that doesn’t appear to be influencing the allele frequencies of the populations.

(1 mark)

*1 Natural selection*

* 1. Describe how the process you mentioned in Question 6 affects the genotype frequencies of populations in general.

(2 marks)

*1 Favourable alleles are passed on as they live to reproduce*

*1 Alleles that are detrimental to survival do not get passed on*

Follow through

Migration:

1 – gene flow of alleles from one population to another

1 – introduction of new alleles to populations (which didn’t have this allele before)

Any 2 of the following:

Speciation:

1 – Two populations separated for an extended period of time with different allele frequencies

1 – Genotype frequencies of populations vary due to environmental influences  
1 – Leads to production of a new species with very different genotype frequencies to the original population

Variation/Mutation/Barrier to gene flow/Random genetic drift would **definitely** affect the population so no follow through marks for these.

1. For the diseases listed below, describe the effect they have on the population, the processes that maintain them in the population AND why it may be of benefit:
   1. Sickle cell anaemia (3 marks)

*1 Homozygous have reduced lifespan so do not pass on*

*1 Heterozygous are protected against malaria*

*1 These individuals therefore survive to pass on the trait*

* 1. Thalassemia (3 marks)

*1 Not generally life threatning OR mild, moderate or severe symptoms depening on the number of genes the individual inherits (up to 4 genes)*

*1 Gene pool isolated due to marriage between cousins along the Mediteranean sea*

*1 Heterozygous carrier protected against malaria*

* 1. Tay Sach’s Disease (chronic form) (3 marks)

*1 Homozygous for Tay Sach’s is fatal in the chronic form*

*1 Genetic drift – small population isolated by religion/language*

*1 Protection against TB if Heterozygous*

**Take home booklet:**

**1 All data in tables correct (see answers below, correct to 1 dp)**

**1 Other questions completed**

**1 Completed A3 sheet**

**References:   
1 3 references in *attempted* APA 6th edition format.**

**1 At least 3 plus references in correct APA 6th edition format.**

**Take home part answers:**

1. Record the following in the tables provided:

a) Count the allele frequencies in both populations.

b) Count the number of allele combinations in both populations.

c) Calculate the allele frequencies as percentages for both, round to 1 decimal place.

|  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- |
| **Original population** | | **Number counted** | **%** |  | **Island population** | | **Number counted** | **%** |
| **Allele types /84** | IA | 33 | 39.3 |  | **Allele types**  **/34** | IA | 16 | 47.1 |
| IB | 10 | 11.9 |  | IB | 0 | 0 |
| i | 41 | 48.8 |  | i | 18 | 52.9 |
| **Allele combinations /42** | IAIA | 9 | 21.4 |  | **Allele combinations**  **/17** | IAIA | 3 | 17.7 |
| IAi | 10 | 23.8 |  | IAi | 10 | 58.8 |
| IBIB | 0 | 0 |  | IBIB | 0 | 0 |
| IBi | 5 | 11.9 |  | IBi | 0 | 0 |
| IAIB | 5 | 11.9 |  | IAIB | 0 | 0 |
| ii | 13 | 31.0 |  | ii | 4 | 23.5 |

|  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- |
| **Original phenotype frequencies** | | | |  | **Island phenotype frequencies** | | | |
| **Type A** | **Type B** | **Type AB** | **Type O** |  | **Type A** | **Type B** | **Type AB** | **Type O** |
| 45.2 | 11.9 | 11.9 | 31.0 |  | 76.5 | 0 | 0 | 23.5 |

2. Use the following formula to calculate a gene (allele) frequency:

**Gene (allele) frequency =**

**number of specific alleles ÷ total number of alleles x 100**

*NOTE: round to 1 decimal place*